NEWBORN SCREENING NEWS

The California Newborn Screening Program **Summer 1997**

Hemoglobin Surveys Provide Insight into Program

The Program

Since 1990, California has screened all newborns for sickle cell disease and other related hemoglobinopathies. In that time, approximately 750 cases of sickle cell disease and other clinically significant hemoglobinopathies have been identified. In addition, each year over 8,000 babies with trait, the majority being sickle cell trait, are detected. Although most hemoglobin traits are associated with few or no medical problems, carrier detection is valuable in that it provides the opportunity to educate families, to test family members and to provide referral to genetic counseling for families at risk for a clinically significant hemoglobinopathy in future pregnancies.

For these reasons, and because of the high frequency and severity of sickle cell disease, the State has developed a Hemoglobin Trait Follow-Up component to the Newborn Screening Program and funds regional Sickle Cell Counseling Centers (SCCCs) to serve as a resource to health care providers. The SCCCs provide free voluntary counseling and testing for families of infants identified with hemoglobin S (sickle cell) trait, D trait or C trait. The SCCCs provide these services at 30 sites throughout California.

The Surveys

In July of 1995, two surveys were designed and administered to evaluate the effectiveness and determine user satisfaction of the SCCCs in providing the following services:

- Trait follow-up counseling and services, including family testing
- Professional education workshops targeting pediatric care providers
- Public education

Two separate surveys, one for providers and the other for parents, were sent out. The parent survey was randomly sent to 2,997 mothers who had a newborn identified with S, C or D trait in 1994. This sample represented approximately half of all S, C and D traits identified that year. The provider survey was sent to 1,650 pediatric care providers who were the physician of record for at least one infant identified with trait (S, C, or D) in 1994. This represented about half of all pediatric care providers within California.

The Results

The parent survey had a response rate of 11.8% (278). A small number of the surveys were completed via telephone call. Although the sample size was small, it appeared to be representative of the total trait population both demographically and geographically.

Of those who responded, 83.5% remembered receiving the initial trait letter following the newborn screen. Of these, 58.2% recalled discussing the letter with their pediatrician and 24.5% had called a SCCC to receive information and counseling. About half of those individuals who called made appointments to go to the Center nearest them.

Those parents who received counseling either by phone or in person from a SCCC were very satisfied with the interaction. Respondents gave 'Excellent' or 'Very Good' scores totaling 74% for phone counseling and 80% for in-person counseling.

However, the majority of parents did not receive counseling from a SCCC. Many reported that they did

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Newborn Screening Conference:
Past Accomplishments and Future Possibilities

October 8-9, 1997
Los Angeles Hyatt Regency

For more info, call the Newborn Screening Section (510) 540-2534

Report on the Sickle Cell Disease and Thalassemias Conference

Mary Brown, Executive Director,
Sickle Cell Disease Research Foundation

The Sickle Cell Disease and Thalassemias Conference was held June 28-29, 1996 in Pasadena, CA. It was co-sponsored by the Pacific Southwest Regional Genetics Network (PSRGN), the Sickle Cell Disease Research Foundation, and the California Department of Health Services.

The meeting aimed to bring together national experts and consumers from the PSRGN region of California, Nevada and Hawaii to learn about the latest research, standards of care at the hospital and community levels, and screening programs in sickle cell disease and thalassemia. The conference also provided a forum to identify gaps in service, assess the impact of managed care, and develop recommendations to address unmet needs.

The conference was heralded as a huge success. It was attended by physicians, nurses, public health and school nurses, psychologists, social workers, genetic counselors, health educators, other professionals, and people with sickle cell disease and their parents. About 150 people attended each day.

The conference featured outstanding speakers and workshops. One of the most critical components was bringing professionals from a variety of settings, including tertiary care centers, private practices, clinics, community agencies, public health departments, along with consumers, together for open dialogue, networking and exchange of information.

CDC Sickle Cell Cooperative Agreement

The Genetic Disease Branch has undertaken a Cooperative Agreement with the Centers for Disease Control, Birth Defect/Genetics Unit to assess risk factors for mortality and morbidity in infants diagnosed with sickle cell disease through Newborn Screening. Two other states, Illinois and New York, are also participating in this cooperative study.

The cohort for the study is infants born in 1992 and 1993 in the three states who were diagnosed with sickle cell disease in the newborn period. All types of sickle cell disease are being included in the study. Each child will be followed until age three. The aim is to determine which risk factors may affect outcome in these children as measured by mortality and frequency and severity of sickle cell disease-related hospitalizations. Biological risk factors will include type of sickle cell disease and Hb S haplotype. Non-biological factors include demographic factors of the infants' mothers (age, education, etc.), location of treatment, type of treatment, penicillin compliance, and parental education.

The three year project has a budget of about $60,000 per year. A research associate, Rhonda Choi, has been hired on the grant to spearhead the efforts. She is an employee of the California Public Health Foundation.

At the time of printing, mortality statistics indicate that only one child in the 1992-93 cohort died from sickle cell disease-related causes by age three. A second child from this birth year died during the fourth year of age. The Genetic Disease Branch has recorded a total of 6 deaths from sickle cell-related causes since screening began in 1990.

Anyone having information on deaths due to sickle cell disease in infants born since 1990 in California is requested to contact Rhonda Choi at (510) 849-5088 or Corinna Tempelis, MPH at (510) 540-3299. Thank you.

Regional Sickle Cell Counseling Centers

Sickle Cell Disease Research Foundation Counseling Center
510 Goldleaf Circle, Suite 150, Los Angeles, CA 90056
(213) 299-3600
Satellite sites: Pasadena (818) 405-0072, San Fernando (818) 837-1955, Santa Barbara (805) 563-9996, Ventura (805) 520-6435

Children's Hospital Oakland, Sickle Cell Counseling Center
747 - 52nd Street, Oakland, CA 94609
(510) 428-3452 or (800) 675-6599
Satellite sites: San Francisco, Richmond, Oakland, Hayward, Fremont, Plesasanton

Sickle Cell Organization of the Inland Counties
2060 University Avenue, Room 206, Riverside, CA 92507
(909) 684-0420 (Riverside, San Bernardino)
Satellite sites: Ontario, Perris, El Centro and Orange County (800) 992-6722

San Diego Sickle Cell Counseling Center
San Diego, CA
(800) 992-6722

Valley Children's Hospital, Sickle Cell Program
3151 North Millbrook, Fresno, CA 93703
(209) 225-3000 ext. 1750
Satellite sites: Bakersfield, Merced (800) 548-5435 ext. 1750

Santa Clara County Health Department
976 Lenzen Avenue, San Jose, CA 94805
(408) 299-5850 or (800) 464-6355
Satellite sites: San Jose, East Palo Alto, Modesto, Monterey/Salinas (800) 464-6355

UC Davis Medical Center, Sickle Cell Counseling Center
2516 Stockton Blvd., Sacramento, CA 95817
(916) 734-2782
Satellite site: Stockton

Sickle Cell Counseling Centers Services Expanded

The State-funded Sickle Cell Counseling Centers are in the process of expanding their services to include a limited range of support services to immediate families of children identified by the Newborn Screening Program with a clinically-significant hemoglobinopathy (initial screening pattern of FS, FSC/FCS, FSD/FDS, FSE/FES, Fsa, FSV/FVS, or F only).

The support services include assistance with obtaining confirmatory testing, health education, sickle cell counseling, transportation, interpretation for medical appointments, and assistance with referrals for genetic counseling and social services. Services will be provided for families of children with initial patterns consistent with sickle cell disease. Services must be requested by primary care providers, clinics, Area Genetic Center NBS Follow-up Coordinators, or CCS Sickle Cell Disease Centers.

Assistance with initial NBS patterns of F only and FE will be limited to confirmatory testing upon request from a NBS Follow-up Coordinator.

Sickle Cell Counseling Centers are listed on page 2 in this newsletter. Services may vary by region based upon need and resources. For more information contact the Sickle Cell Counseling Center in your area.

GeneHELP Resource Center

Spotlight on new materials

The Best Defense: Giving Penicillin to Children with Sickle Cell Disease is a 20 minute videotape for families of young children with sickle cell disease. The production quality and new graphics are very good. We highly recommend this video. CCS Sickle Cell Disease Centers and State-funded Sickle Cell Counseling Centers can get a copy free from GeneHELP. Other agencies can call the New England Research Institute at 1(800) 775-6374 ext. 560.

Management and Therapy of Sickle Cell Disease was revised in 1995 by the US Department of Health and Human Services to reflect the remarkable advances in the treatment of sickle cell disease since the previous edition came out five years earlier. This is an invaluable resource for practitioners. CCS Sickle Cell Disease Centers and State-funded Sickle Cell Counseling Centers can get free copies from GeneHELP. For large quantities, call the National Heart, Lung and Blood Institute at (301) 435-0055.

Call the GeneHELP Resource Center, (510) 540-2534, to find out about other free educational materials.

Kudos! Kudos! Kudos!

Over three hundred hospitals provide perinatal services and six free-standing children's hospitals provide neonatal services in California. It is imperative that the staff at these facilities perform newborn screening procedures correctly, and understand that accurately recording demographic information on the Newborn Screening collection form (NBS-I) is just as important as collecting the specimen.

Each year every hospital receives the Hospital Evaluation Performance Profile (HEPP) report, a quality assurance tool used to improve the process of newborn screening by providing feedback to facilities on their performance. It reflects outliers, e.g. missing data elements, reasons specimens were deemed inadequate, specimens collected too early or too late, and delays in specimen transport. The report enables hospitals to compare their performances with state averages.

For example, if 8% of a hospital's submitted specimen collection forms are missing time of birth, and the statewide facility average for the omission is 5%, then the HEPP report identifies this as a deficiency for the hospital. The report also shows the percentage of babies tested under 12 hours of age, infants tested after transfusion, and other practices that may adversely affect the tests' validity. With this feedback, the staff in every hospital can strive toward improving their procedures.

We recognize that the process of specimen and data collection takes time, teamwork, and patience.

The Newborn Screening Section of the Genetic Disease Branch acknowledges and bestows “balloons of thanks” to the following hospitals that have **zero deficiencies** on the HEPP report for 1996. Note: Hospitals that had fewer than 10 specimen collection forms are not included. The numbers in parentheses indicate the number of newborn screening specimens collected in each hospital for the year, and R = regular, N = NICU.

Antelope Valley Medical Center (4583, R)
Holy Cross Hospital (34, N)
Kaiser, Panorama City (131, N)
Kaiser, Riverside (151, N)
Kaiser, Woodland Hills (217, N)
Lakewood Regional Medical Center (628, R)
Los Alamitos General Hospital (944, R)
Medical Center Garden Grove (173, N)
Mercy Medical Center, Redding (157, N)
Santa Ana Hospital (2364, R)
Woodland Memorial Hospital (17, N)

Hemoglobin Surveys Provide Insight into Program (from page 1)

not call or go to a Center because their pediatrician told them the baby was fine or because the pediatrician counseled them about the baby's trait. About one-third of the parents did not go because they already knew about sickle cell trait. Many of these respondents reported that family members had already been tested. A smaller percentage said they had difficulty in calling or getting to a Center.

Of those who received counseling from their doctor, 65% reported that 15 minutes or less was spent discussing trait with the family. The doctor satisfaction rating was lower than the SCCCs; 41% gave 'Excellent' or 'Very Good' scores. And while most parents remember the physician's staff providing basic information about the genetics of sickle cell, the impact on the child's health, and future pregnancy concerns, few recall receiving information about the SCCC's services, the location of the nearest Center, or that carrier testing can be done directly in the doctor's office. It is a goal of the Newborn Screening Program to encourage providers to share this important information with their patients.

The provider survey gave further insight into many of the issues raised by the parent survey. About 21.4% (343) of the surveys were completed and returned. Respondents stated that over 75% of them provided general information about trait to parents of children identified with trait, and 67% currently refer families to a SCCC. However, only one in three pediatricians reported counseling trait families about the risks for future pregnancies or providing family testing to them.

Only one in five providers reported distributing educational materials to families with trait, but many providers were interested in receiving such materials and using them.

Of those doctors who had referred at least one family to a SCCC, 80% felt that the families were able to understand key concepts and an equal number reported being 'Satisfied' or 'Very Satisfied' with the Center's services. This corresponded well with the satisfaction rates from the patients who received SCCC services.

Two areas that need further examination are follow-up reports and professional education. Only 43.8% of pediatricians responding said they receive follow-up reports on patients who contacted a SCCC. Also, only 10.8% of those responding had attended or had a staff member attend a SCCC workshop.

The Conclusions

While patient satisfaction with the SCCCs was very high, it is hoped that more families will avail themselves of this useful service in the future. These surveys have opened a floodgate of ideas about modifying the way services are marketed and provided. The staff of the Department of Health Services, Newborn Screening Program, in conjunction with the staff at the SCCCs, are committed to providing primary care providers with follow-up reports, educational materials, and workshops on sickle cell trait and hemoglobin disorders. Your continued support of the Newborn Screening Program is appreciated.

If you have any suggestions or questions please write to Kathleen Velazquez, MPH, Chief, Newborn Screening Section or George C. Cunningham, MD, MPH, Chief, Ger Disease Branch -- or call them at (510) 540-2534.

Newborn Screening Coordinators at Area Genetic Centers (AGCs)

UC San Francisco	(415) 476-5048
UC Davis	(916) 754-5400
Children's Hospital, Oakland	(510) 428-3127
Stanford University	(415) 723-7987
Fresno Valley Children's Hospital	(209) 246-6416
Loma Linda University	(909) 824-4191
UC Los Angeles	(310) 825-9719
Children's Hospital, Los Angeles	(213) 669-2226
Harbor/UCLA Medical Center	(310) 222-3751
UC Irvine Medical Center	(714) 456-6878
San Diego-Imperial Counties Developmental Services	(619) 576-2975
Kaiser-Permanente, Northern CA	(510) 596-6192
Kaiser-Permanente, Southern CA	(818) 564-3326

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